

# Rosa J Torres

## List of Publications by Year in descending order

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98  
papers

2,542  
citations

236925

25  
h-index

214800

47  
g-index

108  
all docs

108  
docs citations

108  
times ranked

2148  
citing authors

#	ARTICLE	IF	CITATIONS
1	Delineation of the motor disorder of Lesch-Nyhan disease. <i>Brain</i> , 2006, 129, 1201-1217.	7.6	247
2	Hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency: Lesch-Nyhan syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2007, 2, 48.	2.7	224
3	Diagnosis of gout in patients with asymptomatic hyperuricaemia: a pilot ultrasound study: Table 1. <i>Annals of the Rheumatic Diseases</i> , 2012, 71, 157-158.	0.9	163
4	Attenuated variants of Lesch-Nyhan disease. <i>Brain</i> , 2010, 133, 671-689.	7.6	147
5	Asymptomatic Hyperuricemia: Impact of Ultrasonography. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 592-595.	1.1	127
6	The Spectrum of Hypoxanthine-Guanine Phosphoribosyltransferase (HPRT) Deficiency. <i>Medicine (United Tj ETQq0 0.0 rgBT /Overlock 10</i>	1.0	108
7	Genotype-phenotype correlations in neurogenetics: Lesch-Nyhan disease as a model disorder. <i>Brain</i> , 2014, 137, 1282-1303.	7.6	105
8	UROMODULIN Mutations Cause Familial Juvenile Hyperuricemic Nephropathy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 1398-1401.	3.6	99
9	Update on the Phenotypic Spectrum of Lesch-Nyhan Disease and its Attenuated Variants. <i>Current Rheumatology Reports</i> , 2012, 14, 189-194.	4.7	61
10	Prevalence of left ventricular hypertrophy in patients with mild hypertension in primary care: impact of echocardiography on cardiovascular risk stratification. <i>American Journal of Hypertension</i> , 2003, 16, 556-563.	2.0	51
11	Efficacy and safety of allopurinol in patients with hypoxanthine-guanine phosphoribosyltransferase deficiency. <i>Metabolism: Clinical and Experimental</i> , 2007, 56, 1179-1186.	3.4	47
12	Uric acid excretion in healthy subjects: a nomogram to assess the mechanisms underlying purine metabolic disorders. <i>Metabolism: Clinical and Experimental</i> , 2012, 61, 512-518.	3.4	46
13	Metabolic syndrome: prevalence, associated factors, and C-reactive protein. <i>Metabolism: Clinical and Experimental</i> , 2008, 57, 1232-1240.	3.4	43
14	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in thirteen Spanish families. <i>Human Mutation</i> , 2000, 15, 383-383.	2.5	35
15	Efficacy of Rasburicase in Hyperuricemia Secondary to Lesch-Nyhan Syndrome. <i>American Journal of Kidney Diseases</i> , 2009, 53, 677-680.	1.9	33
16	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 190.	2.7	31
17	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. <i>Kidney International Reports</i> , 2020, 5, 1472-1485.	0.8	30
18	Systematic genetic analysis of early-onset gout: ABCG2 is the only associated locus. <i>Rheumatology</i> , 2020, 59, 2544-2549.	1.9	30

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19	Phosphoribosylpyrophosphate synthetase overactivity as a cause of uric acid overproduction in a young woman. <i>Arthritis and Rheumatism</i> , 2003, 48, 2036-2041.	6.7	29
20	Losartan improves resistance artery lesions and prevents CTGF and TGF- $\beta$ <sup>2</sup> production in mild hypertensive patients. <i>Kidney International</i> , 2006, 69, 1237-1244.	5.2	29
21	Serum Urate, Metabolic Syndrome, and Cardiovascular Risk Factors. A Population-Based Study. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 620-623.	1.1	29
22	Tubular Urate Transporter Gene Polymorphisms Differentiate Patients with Gout Who Have Normal and Decreased Urinary Uric Acid Excretion. <i>Journal of Rheumatology</i> , 2014, 41, 1863-1870.	2.0	29
23	Genetic Mapping Studies of Familial Juvenile Hyperuricemic Nephropathy on Chromosome 16p11-p13. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 464-470.	3.6	26
24	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 427-437.	1.6	26
25	Clinical usefulness of serum tartrate-resistant acid phosphatase in paget's disease of bone: Correlation with other biochemical markers of bone demodelling. <i>Calcified Tissue International</i> , 1991, 49, 14-16.	3.1	25
26	Familial juvenile hyperuricaemic nephropathy (FJHN): linkage analysis in 15 families, physical and transcriptional characterisation of the FJHN critical region on chromosome 16p11.2 and the analysis of seven candidate genes. <i>European Journal of Human Genetics</i> , 2003, 11, 145-154.	2.8	25
27	Genome-wide study of familial juvenile hyperuricaemic (gouty) nephropathy (FJHN) indicates a new locus, FJHN3, linked to chromosome 2p22.1-p21. <i>Human Genetics</i> , 2011, 129, 51-58.	3.8	25
28	Normal HPRT coding region in complete and partial HPRT deficiency. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 167-172.	1.1	24
29	Hypoxanthine deregulates genes involved in early neuronal development. Implications in Lesch-Nyhan disease pathogenesis. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1109-1118.	3.6	23
30	A double-blind, placebo-controlled, crossover trial of the selective dopamine D1 receptor antagonist ecopipam in patients with Lesch-Nyhan disease. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 160-166.	1.1	23
31	Adenosine, dopamine and serotonin receptors imbalance in lymphocytes of Lesch-Nyhan patients. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1129-1135.	3.6	22
32	Mechanisms for phenotypic variation in Lesch-Nyhan disease and its variants. <i>Human Genetics</i> , 2011, 129, 71-78.	3.8	21
33	Consequences of Delayed Dental Extraction in Lesch-Nyhan Disease. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 225-229.	1.5	19
34	Uric Acid Metabolism in Patients with Primary Gout and the Metabolic Syndrome. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2010, 29, 330-334.	1.1	18
35	Levodopa therapy in a Lesch-Nyhan disease patient: Pathological, biochemical, neuroimaging, and therapeutic remarks. <i>Movement Disorders</i> , 2008, 23, 1297-1300.	3.9	17
36	Abnormal adenosine and dopamine receptor expression in lymphocytes of Lesch-Nyhan patients. <i>Brain, Behavior, and Immunity</i> , 2009, 23, 1125-1131.	4.1	17

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37	Metabolic Syndrome in Primary Gout. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2014, 33, 185-191.	1.1	17
38	Purine metabolism in women with primary gout. <i>American Journal of Medicine</i> , 1994, 97, 332-338.	1.5	16
39	Efficacy and Safety of Allopurinol in Patients with the Lesch-Nyhan Syndrome and Partial Hypoxanthine- Phosphoribosyltransferase Deficiency: a Follow-up Study of 18 Spanish Patients. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1077-1082.	1.1	15
40	Hypoxanthine decreases equilibrative type of adenosine transport in lymphocytes from Lesch-Nyhan patients. <i>European Journal of Clinical Investigation</i> , 2007, 37, 905-911.	3.4	15
41	Arts syndrome with a novel missense mutation in the PRPS1 gene: A case report. <i>Brain and Development</i> , 2016, 38, 954-958.	1.1	15
42	Adenosine transport in peripheral blood lymphocytes from Lesch-Nyhan patients. <i>Biochemical Journal</i> , 2004, 377, 733-739.	3.7	14
43	Resolution of Massive Tophaceous Gout with Three Urate-lowering Drugs. <i>American Journal of Medicine</i> , 2013, 126, e9-e10.	1.5	14
44	Skewed X inactivation in Lesch-Nyhan disease carrier females. <i>Journal of Human Genetics</i> , 2017, 62, 1079-1083.	2.3	14
45	Macrocytic anemia in Lesch-Nyhan disease and its variants. <i>Genetics in Medicine</i> , 2019, 21, 353-360.	2.4	14
46	Purine metabolism in female heterozygotes for hypoxanthine-guanine phosphoribosyltransferase deficiency. <i>European Journal of Clinical Investigation</i> , 1998, 28, 950-957.	3.4	13
47	Ultrasonography in the diagnosis of asymptomatic hyperuricemia and gout. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2016, 35, 517-523.	1.1	13
48	A review of the implication of hypoxanthine excess in the physiopathology of Lesch-Nyhan disease. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2016, 35, 507-516.	1.1	13
49	Physiological levels of folic acid reveal purine alterations in Lesch-Nyhan disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 12071-12079.	7.1	13
50	A simplified method for the determination of phosphoribosylpyrophosphate synthetase activity in hemolysates. <i>Clinica Chimica Acta</i> , 1994, 224, 55-63.	1.1	12
51	Metabolic Syndrome Characteristics in Gout Patients. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2010, 29, 325-329.	1.1	12
52	Clinical utility gene card for: Lesch-Nyhan Syndrome - update 2013. <i>European Journal of Human Genetics</i> , 2013, 21, 1187-1187.	2.8	11
53	The Diagnosis Of HPRT Deficiency in the 21st Century. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 564-569.	1.1	10
54	Carrier and prenatal diagnosis of Lesch-Nyhan disease due to a defect in HPRT gene expression regulation. <i>Gene</i> , 2012, 511, 306-307.	2.2	9

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55	HPRT Deficiency in Spain: What Have We Learned in the Past 30 Years (1984–2013)? Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 223-232.	1.1	9
56	The Spectrum of Hypoxanthine-Guanine Phosphoribosyltransferase (HPRT) Deficiency. , 2000, 486, 15-21.		8
57	Familial Nephropathy Associated with Hyperuricemia in Spain: Our Experience with 3 Families Harboring a UMOD Mutation. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1295-1300.	1.1	8
58	Partial HPRT Deficiency Phenotype and Incomplete Splicing Mutation. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 295-300.	1.1	8
59	Efficacy and Safety of a Urate Lowering Regimen in Primary Gout. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 174-180.	1.1	8
60	Novel Developments in Metabolic Disorders of Purine and Pyrimidine Metabolism and Therapeutic Applications of Their Analogs. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 165-173.	1.1	8
61	Methylation Status of HPRT1 Promoter in HPRT Deficiency with Normal Coding Region. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 301-305.	1.1	7
62	Clinical utility gene card for: Lesch–Nyhan syndrome. European Journal of Human Genetics, 2011, 19, 2-3.	2.8	7
63	Fenofibrate Increases Serum Creatinine in a Patient with Familial Nephropathy Associated to Hyperuricemia. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 181-184.	1.1	7
64	<scp>GLUT</scp>9 influences uric acid concentration in patients with Lesch–Nyhan disease. International Journal of Rheumatic Diseases, 2018, 21, 1270-1276.	1.9	7
65	Development of new forms of self-injurious behavior following total dental extraction in Lesch–Nyhan disease. Nucleosides, Nucleotides and Nucleic Acids, 2016, 35, 524-528.	1.1	6
66	La ecografía en el diagnóstico de la hiperuricemia asintomática y la gota. Revista Clinica Espanola, 2016, 216, 445-450.	0.6	6
67	Determination of phosphoribosylpyrophosphate synthetase activity in human cells by a non-isotopic, one step method. Clinica Chimica Acta, 1996, 245, 105-112.	1.1	5
68	Familial juvenile hyperuricaemic nephropathy. QJM - Monthly Journal of the Association of Physicians, 2004, 97, 457-458.	0.5	5
69	The Pathophysiology of Hyperuricemia in Essential Hypertension: A Pilot Study. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1197-1199.	1.1	5
70	Absence of SLC22A12/URAT1 Gene Mutations in Patients with Primary Gout. Journal of Rheumatology, 2012, 39, 1901-1901.	2.0	5
71	Unapparent hypoxanthine-guanine phosphoribosyltransferase deficiency. Clinica Chimica Acta, 2017, 472, 136-138.	1.1	5
72	Toll-Like receptor 4 (TLR4) polymorphism rs2149356 and risk of gout in a Spanish cohort. Nucleosides, Nucleotides and Nucleic Acids, 2020, 39, 1424-1431.	1.1	5

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73	Lesch-Nyhan syndrome. <i>Drugs of the Future</i> , 2010, 35, 421.	0.1	5
74	Binding of serum osteocalcin to hydroxyapatite in Paget's disease of bone. <i>Bone and Mineral</i> , 1991, 14, 55-65.	1.9	4
75	Osseous and Intestinal Compartments in the Humoral Hypercalcemia of Malignancy Associated to Walker 256 Tumor in Rats. <i>Oncology</i> , 1993, 50, 148-152.	1.9	4
76	Adenosine Transport in HPRT Deficient Lymphocytes from Lesch-Nyhan Disease Patients. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2004, 23, 1193-1196.	1.1	4
77	Hypoxanthine Effects on Cyclic AMP Levels in Human Lymphocytes. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2004, 23, 1181-1183.	1.1	4
78	De la mutación al fenotipo; variabilidad clínica en la enfermedad de Lesch-Nyhan. El papel de la epigenética. <i>Revista Clínica Española</i> , 2014, 214, 461-465.	0.6	4
79	Current understanding of Lesch-Nyhan disease and potential therapeutic targets. <i>Expert Opinion on Orphan Drugs</i> , 2019, 7, 349-361.	0.8	4
80	Effects of Hypoxanthine on Adenosine Transport in Human Lymphocytes. Implications in the Pathogenesis of Lesch-Nyhan Syndrome. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2004, 23, 1177-1179.	1.1	3
81	Hypoxanthine Effect on Equilibrative and Concentrative Adenosine Transport in Human Lymphocytes. Implications in the Pathogenesis of Lesch-Nyhan Syndrome. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2006, 25, 1065-1069.	1.1	3
82	Aicar effect in early neuronal development. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2018, 37, 261-272.	1.1	3
83	Purine Metabolism in Female Patients with Primary Gout. <i>Advances in Experimental Medicine and Biology</i> , 1995, 370, 69-72.	1.6	3
84	Gout New Questions for an Ancient Disease. <i>Advances in Experimental Medicine and Biology</i> , 1998, 431, 1-5.	1.6	3
85	Genetic Diagnosis of Hypoxanthine-Guanine Phosphoribosyltransferase (Hgp <sub>rt</sub> ) Carrier Status by Restriction Analysis and Directed Mutagenesis. <i>Advances in Experimental Medicine and Biology</i> , 1998, 431, 201-204.	1.6	3
86	The trials of alternative medicine. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2003, 96, 172-172.	0.5	2
87	An Unusual Patient with Hypothyroidism, Tophaceous Gout, and Marked Joint Destruction. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 604-607.	1.1	2
88	Enzyme activity and brain anatomy: lessons from HPRT deficiency. <i>Lancet Neurology</i> , The, 2013, 12, 1129-1131.	10.2	2
89	Determination of the Activity of Recombinant Human Phosphoribosylpyrophosphate Synthetase Isoform 1 by a Non-Isotopic, One-Step Method. <i>Advances in Experimental Medicine and Biology</i> , 1995, 370, 821-824.	1.6	2
90	Symptomatic heterozygous X-Linked myotubular myopathy female patient with a large deletion at Xq28 and decrease expression of normal allele. <i>European Journal of Medical Genetics</i> , 2021, 64, 104170.	1.3	1

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91	Safety and Efficacy of Botulinum Toxin in the Treatment of Self-Biting Behavior in Lesch-Nyhan Disease. <i>Pediatric Neurology</i> , 2022, 127, 6-10.	2.1	1
92	Urinary Guanidinoacetate and Creatine Levels in Patients with HPRT Deficiency. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2008, 27, 575-577.	1.1	0
93	R�plica a �«Tratamiento de la hiperuricemia asintom�tica�». <i>Revista Clinica Espanola</i> , 2017, 217, 244.	0.6	0
94	Automedicaci�n en el domicilio frente a lectura de la presi�n arterial en la consulta en el seguimiento de diab�ticos tipo ii : efecto sobre la presi�n arterial ambulatoria y la albuminuria. Estudio aleatorizado. <i>Medicina Cl�nica</i> , 2018, 150, 413-420.	0.6	0
95	Citocinas y marcadores de activaci�n coagulativa en ancianos con fiebre de causa infecciosa. <i>Medicina Cl�nica</i> , 2004, 122, 358-358.	0.6	0
96	An unusual cause of stuttering. , 2008, , 349-351.		0
97	HGPRT and Aprt Activities in Hemolysates During the First Year of Life. <i>Advances in Experimental Medicine and Biology</i> , 1998, 431, 141-145.	1.6	0
98	Hereditary spastic paraplegia associated with a novel homozygous intronic noncanonical splice site variant in the <i>AP4B1</i> gene. <i>Annals of Human Genetics</i> , 2022, 86, 109-118.	0.8	0